Trisomy 13 (Patau syndrome)

What is it?

Trisomy 13, or Patau syndrome, is a congenital chromosomal condition. **Chromosomes** are located inside the cells in our body. Normally, people are born with 23 pairs of chromosomes. Infants born with this syndrome have three copies of chromosome 13 in each cell, whereas most people have only two copies. The additional copies of chromosome 13 can cause physical and intellectual disabilities. Symptoms of trisomy 13 can be severe and life-threatening. Unfortunately, death commonly occurs within the first few days or weeks after birth. Only 5%-10% of infants are still alive after one year. Common symptoms include:

Neurologic disorders (developmental delay, mental retardation)
Craniofacial defects (cleft lip/palate, small head, flat nose, wide-set small eyes, low-set ears)
Congenital heart defects (atrial or ventricular septal defect, PDA, pulmonary artery defect)

How common is it?

About 1 in 16,000 infants born each year will be diagnosed with trisomy 13. A woman's chance of giving birth to an infant with trisomy 13 increases with her age.

What causes it?

Infants born with this syndrome have three copies of chromosome 13 in each cell. The additional copies of chromosome 13 can cause physical and intellectual disabilities. This condition is usually a random event and can happen during the development of the fetus. Inheritance is rare.

How is it treated?

Trisomy 13 is confirmed by chromosomal analysis, a test that looks at and determines the number of chromosomes in a person. Trisomy 13 can be diagnosed during pregnancy by ultrasound, amniocentesis (procedure to obtain amniotic fluid), or chorionic villus sampling (biopsy of the placenta). In addition, infants can be evaluated after birth with a physical exam.

Treatment will vary for each person. It may include medical and physical therapy services. The goal is to improve the overall quality of life. Your child's doctor will discuss appropriate treatment options with you.

For more information:

U.S. National Library of Medicine, Genetics Home Reference
https://medlineplus.gov/genetics/condition/trisomy-13/#inheritance

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/trisomy-13-syndrome/

BDPR Indiana Birth Defects and Problems Registry

Indiana Department of Health

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